Missouri Newborn Screening Disorders Tested and Reported

Classical galactosemia (GALT)
Congenital adrenal hyperplasia (CAH)
Congenital primary hypothyroidism (CH)
Cystic fibrosis (CF)

Amino Acid Disorders

Argininemia (ARG, arginase deficiency)
Argininosuccinate acidemia (ASA, argininosuccinase)
Citrullinemia type I (CIT-I, argininosuccinate synthetase)
Citrullinemia type II (CIT-II, citrin deficiency)
Defects of biopterin cofactor biosynthesis (BIOPT-BS)
Defects of biopterin cofactor regeneration (BIOPT-RG)
Homocystinuria (HCY, cystathionine beta synthase)
Hyperphenylalaninemia (H-PHE)
Hypermethioninemia (MET)
Maple syrup urine disease (MSUD, branched-chain ketoacid dehydrogenase)
Phenylketonuria (PKU, phenylalanine hydroxylase)
Tyrosinemia type I (TYR-1, fumarylacetoacetate hydrolase) *
Tyrosinemia type II (TYR-II, tyrosine aminotransferase)
Tyrosinemia type III (TYR-III, hydroxyphenylpyruvate dioxygenase)

Fatty Acid Disorders

Carnitine acylcarnitine translocase deficiency (CACT)
Carnitine uptake defect (CUD, carnitine transport defect) *
Carnitine palmitoyl transferase deficiency I (CPT-1a)
Carnitine palmitoyl transferase deficiency II (CPT-II)
Dienoyl-CoA reductase deficiency (DE-RED)
Glutaric acidemia type II (GA-II, multiple acyl-CoA dehydrogenase deficiency)
Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
Trifunctional protein deficiency (TFP)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG, SBCAD)
3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
Glutaric acidemia type I (GA-1, glutaryl-CoA dehydrogenase)
Isobutyryl-CoA dehydrogenase deficiency (IBG)

Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase)
Malonic acidemia (MAL, malonyl-CoA decarboxylase)
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)
Methylmalonic acidemia (CBL C,D)
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
Propionic acidemia (PROP, propionyl-CoA carboxylase)

Hemoglobinopathies

Sickle cell disease (Hb S/S)
Sickle hemoglobin-C disease (Hb S/C)
Sickle beta zero thalassemia disease
Sickle beta plus thalassemia disease
Sickle hemoglobin-D disease
Sickle hemoglobin-E disease
Sickle hemoglobin-O-Arab disease
Sickle hemoglobin Lepore Boston disease
Sickle HPFH disorder
Sickle "Unidentified"
Hemoglobin-C beta zero thalassemia disease
Hemoglobin-C beta plus thalassemia disease
Hemoglobin-E beta zero thalassemia disease
Hemoglobin-E beta plus thalassemia disease
Hemoglobin-H disease
Homozygous beta zero thalassemia disease
Homozygous-C disease
Homozygous-E disorder
Double heterozygous beta thalassemia disease

Others

Hearing

* There is a lower probability of detection of this disorder during the immediate newborn period.

The Missouri Newborn Screening Laboratory's goal is to identify infants at risk and in need of diagnostic testing for the above disorders. A normal screening result does **NOT** rule out the possibility of an underlying metabolic/genetic disease.

Reviewed: 10/06/08